



## SAMHD1 gene

SAM and HD domain containing deoxynucleoside triphosphate triphosphohydrolase 1

### Normal Function

The *SAMHD1* gene provides instructions for making a protein whose function is not well understood. The SAMHD1 protein is believed to be involved in the immune system, including the inflammatory process and response to viral infections.

### Health Conditions Related to Genetic Changes

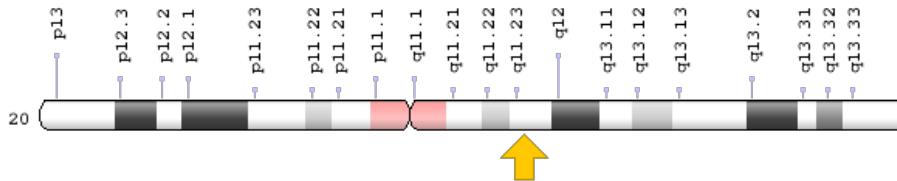
#### Aicardi-Goutieres syndrome

At least 16 mutations in the *SAMHD1* gene have been identified in people with Aicardi-Goutieres syndrome. Mutations in this gene likely result in a SAMHD1 protein that does not function properly. It is not known how this protein dysfunction leads to immune system abnormalities, inflammatory damage to the brain and skin, and other characteristics of Aicardi-Goutieres syndrome.

### Chromosomal Location

Cytogenetic Location: 20q11.23, which is the long (q) arm of chromosome 20 at position 11.23

Molecular Location: base pairs 36,890,882 to 36,951,843 on chromosome 20 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

### Other Names for This Gene

- AGS5
- DCIP
- dendritic cell-derived IFNG-induced protein

- HDDC1
- Mg11
- monocyte protein 5
- MOP-5
- SAM domain and HD domain 1
- SAM domain and HD domain-containing protein 1
- SAMH1\_HUMAN
- SBBI88

## **Additional Information & Resources**

### GeneReviews

- Aicardi-Goutieres Syndrome  
<https://www.ncbi.nlm.nih.gov/books/NBK1475>

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28SAMHD1%5BTIAB%5D%29+OR+%28MOP-5%5BTIAB%5D%29+OR+%28AGS5%5BTIAB%5D%29%29+AND+%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

### OMIM

- SAM DOMAIN- AND HD DOMAIN-CONTAINING PROTEIN 1  
<http://omim.org/entry/606754>

### Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology  
[http://atlasgeneticsoncology.org/Genes/GC\\_SAMHD1.html](http://atlasgeneticsoncology.org/Genes/GC_SAMHD1.html)
- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=SAMHD1%5Bgene%5D>
- HGNC Gene Family: Sterile alpha motif domain containing  
<http://www.genenames.org/cgi-bin/genefamilies/set/760>
- HGNC Gene Symbol Report  
[http://www.genenames.org/cgi-bin/gene\\_symbol\\_report?q=data/hgnc\\_data.php&hgnc\\_id=15925](http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=15925)

- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/25939>
- UniProt  
<http://www.uniprot.org/uniprot/Q9Y3Z3>

## Sources for This Summary

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